



IAFS 2014 *20th World Meeting of the International Association of Forensic Sciences*

**WORLD
FORENSIC
FESTIVAL**

World Forensics Festival
Seoul, Korea
October 15, 2014



International Standards in Forensic DNA

John M. Butler, Ph.D.

National Institute of Standards and Technology
NIST Fellow & Special Assistant to the Director for Forensic Science
Vice-Chair, National Commission on Forensic Science

Definition of “Standard”

stan·dard *noun* \ 'stan-dərd\ :

- a level of quality, achievement, etc., that is considered acceptable or desirable
- something set up and established by authority as a rule for the measure of quantity, weight, extent, value, or quality

Presentation Outline

Why?

- Benefits of standards
 - Help improve quality and consistency in testing

Who?

- Authority to establish standards
 - Expert groups like SWGDAM, ISFG DNA Commission, ENFSI DNA WG, AFSN, OSAC

What?

- Documentary and physical standards

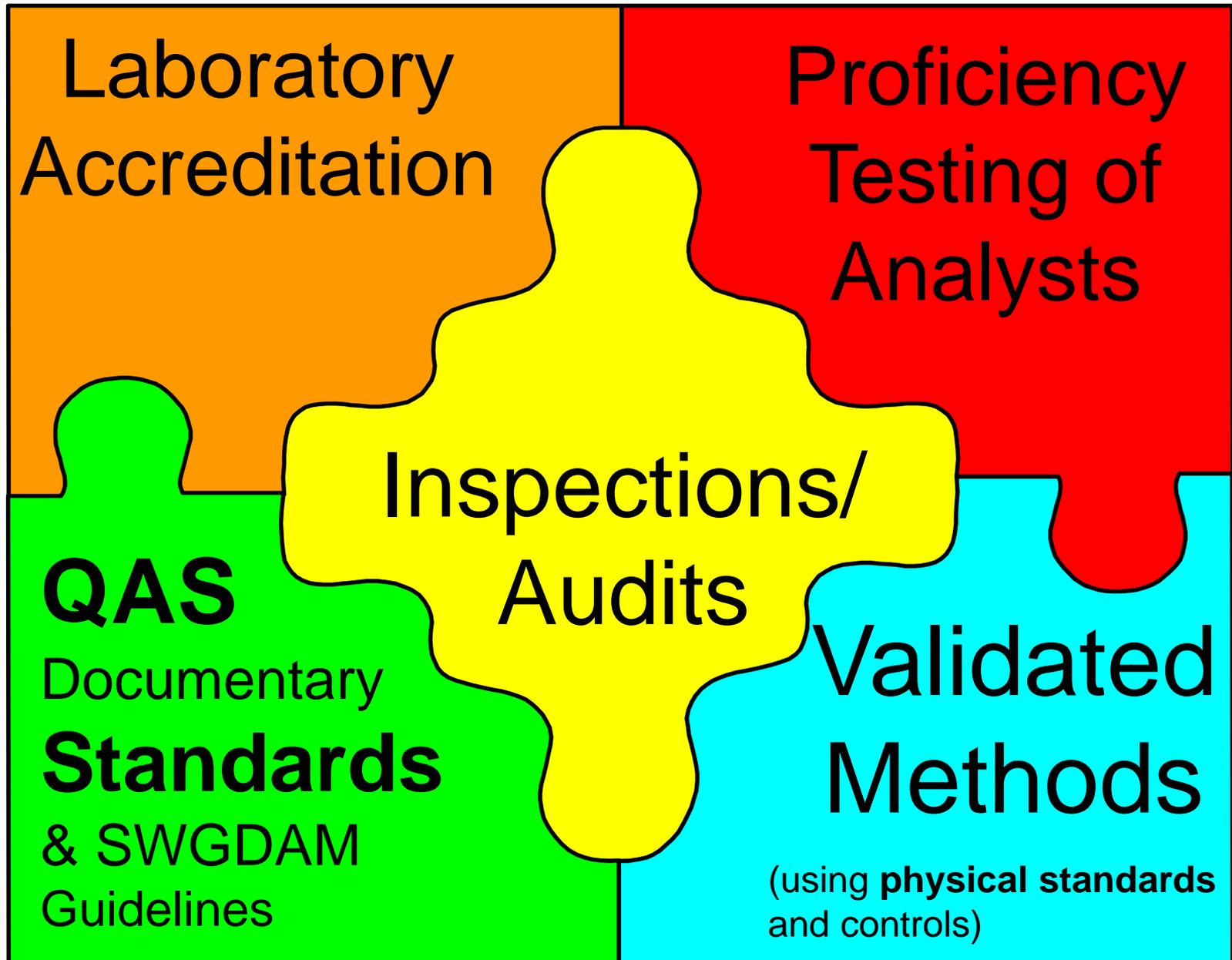
How?

- Quality Assurance Standards (QAS)
- Core loci and common data formats
- Certified Reference Materials (e.g., NIST SRM)

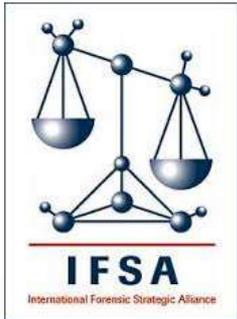
Standards Aid Consistency and Quality

- Consistent processes
 - Documentary standards
- Reliable data comparison
 - Standard data formats and core DNA testing regions
- **I believe that the use of standard methods and materials are a primary reason that DNA is on a more solid foundation compared to many of the other forensic disciplines**

Ensuring Accurate Forensic DNA Results



International and Regional Coordination Efforts in Forensic Science



2004



2008



1968



1974



2004



1995



2008



2008



1986

Organizations Assisting Forensic Science Quality Assurance

Organization; year started	Membership	Website
American Society of Crime Laboratory Directors (ASCLD); started in 1974	U.S. federal, state, and local lab managers; not directly associated with SWGDAM but ASCLD/LAB (not ASCLD) uses the FBI Quality Assurance Standards for DNA audits	http://www.asclcd.org
European Network of Forensic Science Institutes (ENFSI); started in 1995	16 working groups including one on DNA	http://www.enfsi.eu
UK Forensic Science Regulator; started in 2008	Multiple advisory groups inform the Regulator including one on DNA	https://www.gov.uk/government/organisations/forensic-science-regulator
Senior Managers of Australian and New Zealand Forensic Laboratories (SMANZFL); started in 1986	8 Specialist Advisory Groups (SAG) including one on biology (BSAG)	http://www.nifs.com.au/SMANZFL/SMANZFL.html
Academia Iberoamericana de Criminalística y Estudios Forenses (AICEF); started in 2004	Represents 19 Spanish and Portuguese speaking countries in Europe and Latin America; has four working groups including one on forensic genetics	http://www.aicef.net/
Asian Forensic Sciences Network (AFSN); started in 2008	5 working groups including one on DNA	http://www.asianforensic.net

Butler, J.M. (2013) Forensic DNA advisory groups: DAB, SWGDAM, ENFSI, and BSAG. Chapter in Siegel, J.A. & Saukko, P.J. (editors) *Encyclopedia of Forensic Sciences, Second Edition*. Elsevier Academic Press: San Diego. pp. 339-343.



International Forensic Strategic Alliance (IFSA) initiated in Nov 2004

- <http://www.enfsi.eu/ifsa>
- **Crafting minimum requirements documents** to aid developing countries with forensic science
- Released in Oct 2014:
“Minimum Requirements for DNA Collection, Analysis, and Interpretation”

multilateral partnership between the regional networks of operational forensic laboratories



US & Canada



Europe



Australia,
New Zealand



Latin America,
South America,
Spain & Portugal



Asia



South Africa

Forensic DNA Advisory Groups

- ISFG DNA Commission (International)
- FBI DNA Advisory Board (U.S.)
- SWGDAM (U.S.)
- ENFSI DNA WG (Europe)
- Forensic Science Regulator (UK)
- Biology Specialist Advisory Group (Australia/NZ)
- Asian Forensic Science Network DNA WG (Asia)
- **NCFS and OSAC (U.S.)**



Forensic DNA Advisory Groups

Organization	Membership	Meeting Frequency/Purpose
DNA Commission of the International Society for Forensic Genetics (ISFG)	ISFG Executive Committee and selected experts; chaired by Dr. Peter Gill	As needed to prepare recommendations (see http://www.isfg.org/Publications/DNA+Commission)
Scientific Working Group on DNA Analysis Methods (SWGDM)	U.S. and Canada federal, state, and local DNA Technical Leaders and invited guests (40-50 people total); subdivided into 5-8 committees	Meets twice a year to develop guidelines on validation, DNA data interpretation, and other topics
European Network of Forensic Science Institutes (ENFSI) DNA Working Group	>30 European countries and invited guests (90-100 people total); subdivided into 5 committees	Meets twice a year along with European DNA Profiling Group (EDNAP)
Biology Specialist Advisory Group (BSAG)	Representatives of each forensic DNA lab in Australia & New Zealand (11 people total)	Meets once a year under direction of SMANZFL and with support of the Australian National Institute of Forensic Science
Organization of Scientific Area Committees (OSAC)	24 discipline-specific subcommittees (has two DNA groups focused on methods and interpretation)	Just starting in 2014 ; plans to meet once a year in person and multiple times virtually

Butler, J.M. (2013) Forensic DNA advisory groups: DAB, SWGDAM, ENFSI, and BSAG. Chapter in Siegel, J.A. & Saukko, P.J. (editors) *Encyclopedia of Forensic Sciences, Second Edition*. Elsevier Academic Press: San Diego. pp. 339-343.

and recent events at NIST – see <http://www.nist.gov/forensics/osac/index.cfm>



International Society for Forensic Genetics (ISFG) DNA Commission Recommendations

Topics Addressed	Publications (16 as of 2014)
DNA polymorphisms	FSI 1989 (43:109-111) FSI 1992 (52:125-130) FSI 1992 (55:1-3)
Commentary on the 1992 NRC I report	FSI 1993 (59:1-2)
STR markers and allele nomenclature	IJLM 1994 (107:159-160) IJLM 1997 (110:175-176)
Mitochondrial DNA typing Revised and extended guidelines	FSI 2000 (110:79-85) FSIG 2014 (13:134-142)
Y-chromosome STRs Additional recommendations on nomenclature	FSI 2001 (124:5-10) FSI 2006 (157:187-197)
Interpretation of DNA mixtures STR typing results using probabilistic methods	FSI 2006 (160:90-101) FSIG 2012 (6:679-688)
Non-human (animal) DNA	FSIG 2011 (5:501-505)
Disaster victim identification	FSIG 2007 (1:3-12)
Paternity Testing Commission Biostatistics in paternity testing	FSI 2002 (129:148-157) FSIG 2007 (1:223-231)

FSI: *Forensic Science International*; IJLM: *International Journal of Legal Medicine*; FSIG: *Forensic Science International: Genetics*



European Network of Forensic Science Institutes (ENFSI) DNA Working Group Documents Available

One of now 17 working groups in ENFSI; meets at least once each year typically in April; representatives from 35 countries

<http://www.enfsi.eu/about-enfsi/structure/working-groups/dna>

Year	Document
May 2004	Terms and Abbreviations
2009	Report on DNA Legislation in Europe
April 2006	Report on Criminal Cases in Europe Solved by DNA Mass Testing
Nov 2010	Recommended Minimum Criteria for the Validation of Various Aspects of the DNA Profiling Process
Nov 2010	Training DNA staff
Nov 2010	Contamination Prevention Guidelines
Dec 2013	Survey of DNA Databases in Europe
April 2014	DNA Database Management: Review and Recommendations

Selects core DNA testing markers for Europe

Gill, P., et al. (2006a). The evolution of DNA databases-Recommendations for new European STR loci. *Forensic Science International*, 156, 242-244.

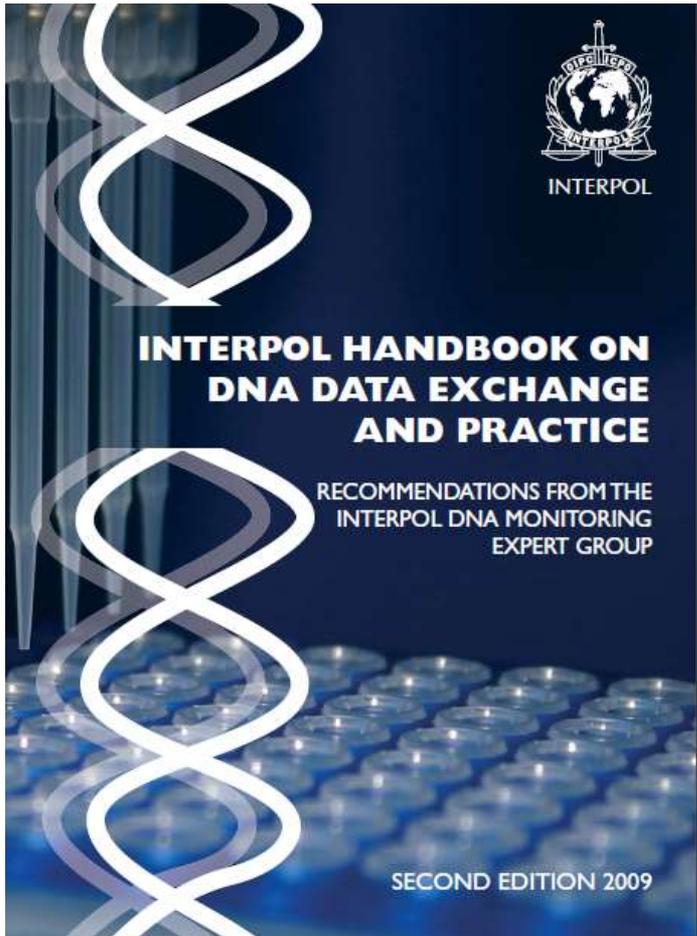
Gill, P., et al. (2006b). New multiplexes for Europe-amendments and clarification of strategic development. *Forensic Science International*, 163, 155-157.



ENFSI DNA Database Management: Review and Recommendations

- Since first version in 2008, this document is **revised each April** by Kees van der Beek from the Netherlands Forensic Institute
- Current document is 88 pages long with **33 recommendations** (and questions for audit purposes)
- Useful and up-to-date information provided on European DNA database activities

Interpol Handbook



118 pages (pdf document)

- Interpol Standard Set of Loci (ISSOL) are the same as the European Standard Set (ESS)
- in 2010, ISSOL was expanded from 7 to 12 loci
- Supports ENFSI DNA Database Management recommendations

<http://www.interpol.int/INTERPOL-expertise/Forensics/DNA>

<https://www.gov.uk/government/organisations/forensic-science-regulator>

Several documents have been published recently

- **Codes of Practice and Conduct** for forensic science providers and practitioners in the Criminal Justice System (Aug 2014, 56 pages)
- FSR-C-108 **APPENDIX: DNA Analysis** (Sept 2014, 11 pages): DNA analysis: codes of practice and conduct
- FSR-G-213 **GUIDANCE** (Sept 2014, 15 pages): **Allele frequency databases and reporting** guidance for the DNA (Short Tandem Repeat) profiling; contains 8 recommendations
- FSR-P-302 **PROTOCOL** (Sept 2014, 49 pages): **DNA contamination detection** -The management and use of staff elimination DNA databases

Concerns have been Raised over Potential for DNA Contamination



June 2014; 100 pages

Previous article by Peter Gill on this topic:

- Gill, P. (1997). The utility of 'substrate controls' in relation to 'contamination'. *Forensic Science International*, 85(2):105-111.
- Gill, P., & Kirkham, A. (2004). Development of a simulation model to assess the impact of contamination in casework using STRs. *Journal of Forensic Sciences*, 49(3): 485-491.
- Gill, P., et al. (2010). Manufacturer contamination of disposable plastic-ware and other reagents—an agreed position statement by ENFSI, SWGDAM and BSAG. *Forensic Science International: Genetics*, 4(4): 269-270.

DNA Contamination Concerns

- *Forensic Sci. Int. Genet.* (July 2010) statement by ENFSI, SWGDAM, and BSAG
- Written to commercial manufacturers of disposable plastic-ware and other reagents used by forensic DNA laboratories worldwide
- Advocates that manufacturers: (1) utilize automation in manufacturing lines, (2) minimize interaction of staff with manufacturing lines, (3) ensure products are protected from staff using personal protective equipment, (4) utilize clean rooms for production, (5) perform QC checks with adequate sensitivity, (6) conduct post-manufacture DNA contaminant destruction, (7) perform QC checks on post-production treatment(s), and (8) maintain staff elimination databases for screening DNA results as needed

Interim Country-Specific Standards Related to DNA Contamination Issues

Guided by the 2010 ENFSI, SWGDAM, and BSAG *FSI Genetics* statement, standards have been developed...

- **British Standard PAS 377:2012** (June 2012, 24 pages):
“Specification for consumables used in the collection, preservation and processing of material for forensic analysis. Requirements for product, manufacturing and forensic kit assembly”
- **Australia Standard AS 5483-2012** (June 2012, 11 pages):
“Minimizing the risk of contamination in products used to collect and analyse biological material for forensic DNA purposes”

ISO/IEC 18385 Standard

- “Minimizing the risk of DNA contamination in products used to collect and analyze biological material for forensic purposes”
- **Under development** and review by participating ISO member countries



International
Organization for
Standardization



International
Electrotechnical
Commission

FBI Quality Assurance Standards (QAS)

- **DNA Identification Act of 1994**
 - Requires FBI Laboratory, those labs receiving federal funds, and those labs using the National DNA Index System (NDIS) to comply
- FBI Laboratory's DNA Advisory Board (**DAB**)
 - Met from 1995 to 2000 to discuss and draft QAS
 - FBI Director issued initial QAS in October 1998 (caseworking) and April 1999 (databasing)
- Scientific Working Group on DNA Analysis Methods (**SWGDM**)
 - assumed responsibility for QAS revisions when DAB was dissolved
 - QAS revisions released in July 2009 and September 2011
- **QAS audit documents are used by accrediting bodies** such as ASCLD/LAB in audits of DNA laboratories as **supplemental material to the ISO/IEC 17025 standard**



The FBI Quality Assurance Standards

Tim Zolandz (FBI) will cover the QAS in more detail later this afternoon

QUALITY ASSURANCE STANDARDS FOR DNA DATABASING LABORATORIES

QUALITY ASSURANCE STANDARDS FOR FORENSIC DNA TESTING LABORATORIES

This document consists of definitions and standards. The standards are quality assurance measures that place specific requirements on the laboratory. Equivalent measures not outlined in this document may also meet the standard if determined sufficient through an accreditation process.

EFFECTIVE DATE:

These standards shall take effect September 1, 2011.

REFERENCES: Federal Bureau of Investigation, "Quality Assurance Standards for Forensic DNA Testing Laboratories" and "Quality Assurance Standards for Convicted Offender DNA Databasing Laboratories," Forensic Science Communications, July 2000, Volume 2, Number 3.

THE FBI QUALITY ASSURANCE STANDARDS

THE FBI QUALITY ASSURANCE STANDARDS

AUDIT FOR

FORENSIC DNA TESTING LABORATORIES

IN ACCORDANCE WITH

THE QUALITY ASSURANCE STANDARDS

FOR

FORENSIC DNA TESTING LABORATORIES

EFFECTIVE SEPTEMBER 1, 2011

1. SCOPE
2. DEFINITIONS
3. QUALITY ASSURANCE PROGRAM
4. ORGANIZATION AND MANAGEMENT
5. PERSONNEL
6. FACILITIES
7. EVIDENCE (*SAMPLE*) CONTROL
8. VALIDATION
9. ANALYTICAL PROCEDURES
10. EQUIPMENT CALIBRATION AND MAINTENANCE
11. REPORTS
12. REVIEW
13. PROFICIENCY TESTING
14. CORRECTIVE ACTION
15. AUDITS
16. SAFETY
17. OUTSOURCING

<http://www.fbi.gov/about-us/lab/biometric-analysis/codis/qas-standards-for-forensic-dna-testing-laboratories-effective-9-1-2011>

<http://www.fbi.gov/about-us/lab/biometric-analysis/codis/qas-standards-for-dna-databasing-laboratories-effective-9-1-2011>



Scientific Working Group on DNA Analysis Methods (SWG-DAM)

<http://www.swgdam.org/>

- **Established in November 1988 by FBI Laboratory**
- Named Technical Working Group on DNA Analysis Methods (TWGDAM) for the first decade
- **Comprised of ~50 scientists from U.S. and Canada**
 - Typically 20-25 voting members and the rest as invited guests
- European Network of Forensic Science Institutes (ENFSI) DNA Working Group representative often attends
- **Three day meetings held semiannually every January and July**
- **Current committees (6) and working groups (2):**
 - Autosomal STR Interpretation, Combined DNA Index System, Enhanced Detection Methods and Interpretation, Quality Assurance, Rapid DNA, Y-STR, **Probabilistic Genotyping**, and **Next Generation Sequencing**
- Previous committees:
 - RFLP, PCR, mitochondrial DNA, mass spectrometry, training, validation, expert systems, missing persons/mass disasters, and mixture interpretation



Current SWGDAM Guidelines

Hyperlinks to documents available on SWGDAM.org

Release Date	Guidelines	Previous Versions (TWGDAM)
2010	<u>STR Interpretation Guidelines</u>	2000
2012	<u>Validation Guidelines for Forensic DNA Analysis Methods</u>	1991, 1995, 2004
2013	<u>Mitochondrial DNA Analysis Interpretation Guidelines</u> & <u>Mitochondrial DNA Nomenclature Example</u>	1993, 2003
2013	<u>Training Guidelines</u>	2001
2014	<u>Guidelines for Missing Persons Casework</u>	--
2014	<u>Interpretation Guidelines for Y-Chromosome STRs</u>	2009

Guidelines in development: enhanced detection methods, validation of probabilistic genotyping software, and updated STR interpretation guidelines

Current Hierarchy of Standards for Accrediting Bodies to Use in Auditing U.S. Forensic DNA Laboratories



International Laboratory Accreditation Cooperation (ILAC)
G19:08/2014 Modules in a Forensic Science Process



**ISO/IEC 17025:2005 General requirements for the
competence of testing and calibration laboratories**

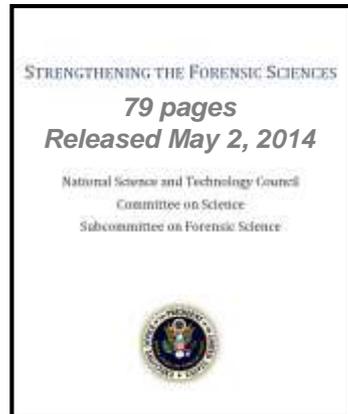
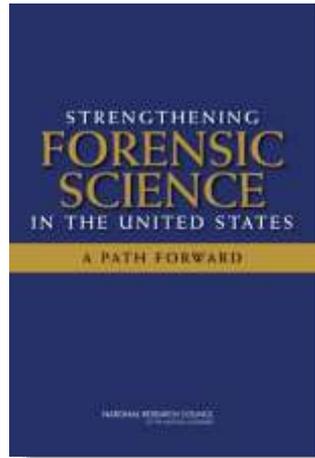


The FBI Quality Assurance Standards (2011) serve as
supplemental materials to ISO/IEC 17025 for DNA audits



SWGDM guidelines (interpretation, validation, etc.)
provide further information but are not audited against

NCFS and OSAC: U.S. Efforts to Strengthen Forensic Science



http://www.whitehouse.gov/sites/default/files/microsites/ostp/NSTC/strengthening_the_forensic_sciences_may_-_2014.pdf

- National Academy of Sciences (**NAS**) **report** issued in Feb 2009
- White House **Subcommittee on Forensic Science** (SoFS) operated from July 2009 to Dec 2012

DOJ/NIST Partnership (announced Feb 2013)

1. **NCFS** (National Commission on Forensic Science)
 - First meeting held February 3-4, 2014 in Washington DC
2. **OSAC** (Organization of Scientific Area Committees)
 - **Being organized**; first public meetings to be held Feb 2015

Department of Justice

Policy focused

Limited Term (FACA)

Attorney General

Recommendations



National Commission
on Forensic Science
(NCFS)

NIST

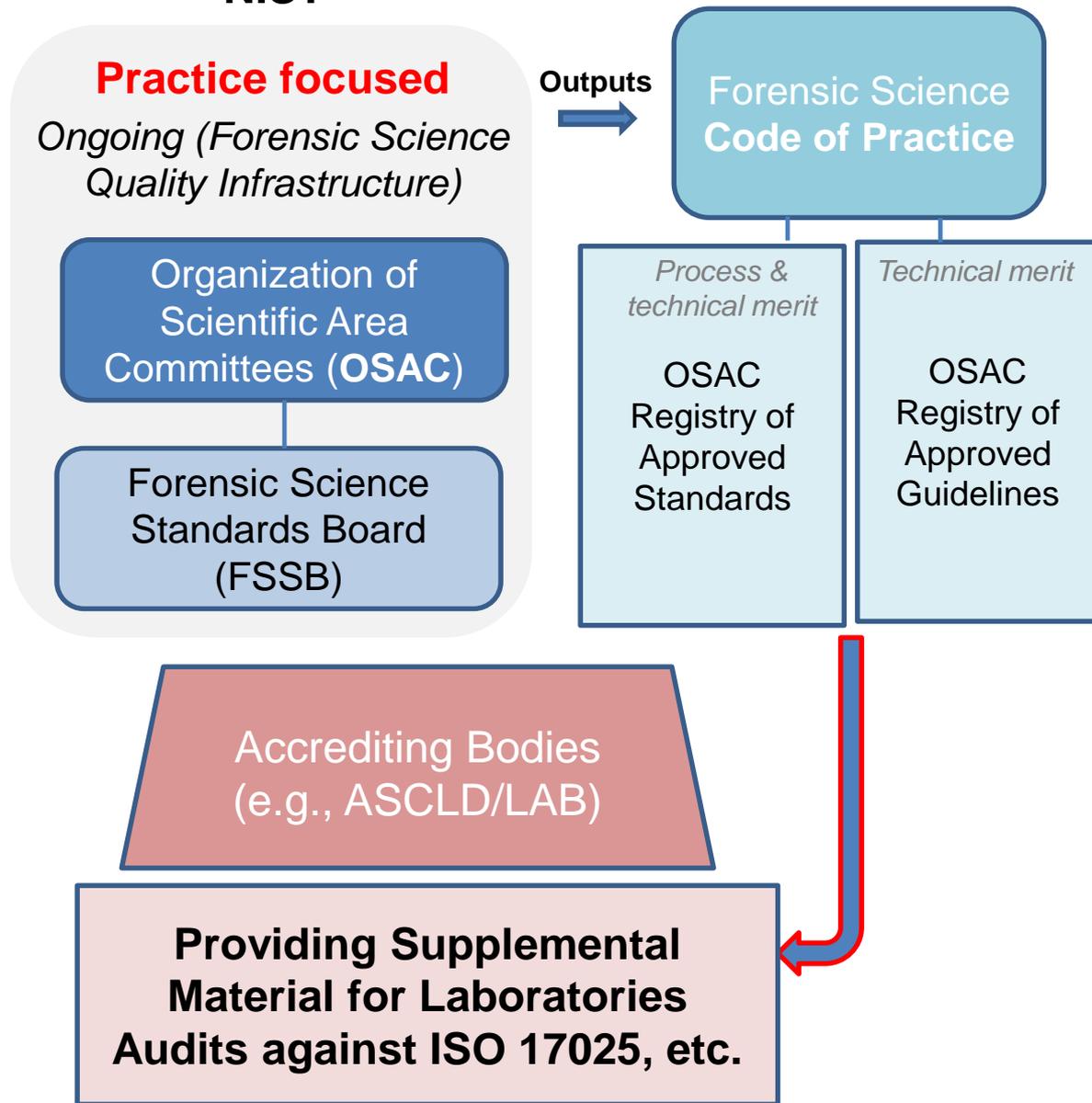
Practice focused

*Ongoing (Forensic Science
Quality Infrastructure)*

Organization of
Scientific Area
Committees (**OSAC**)

Forensic Science
Standards Board
(FSSB)

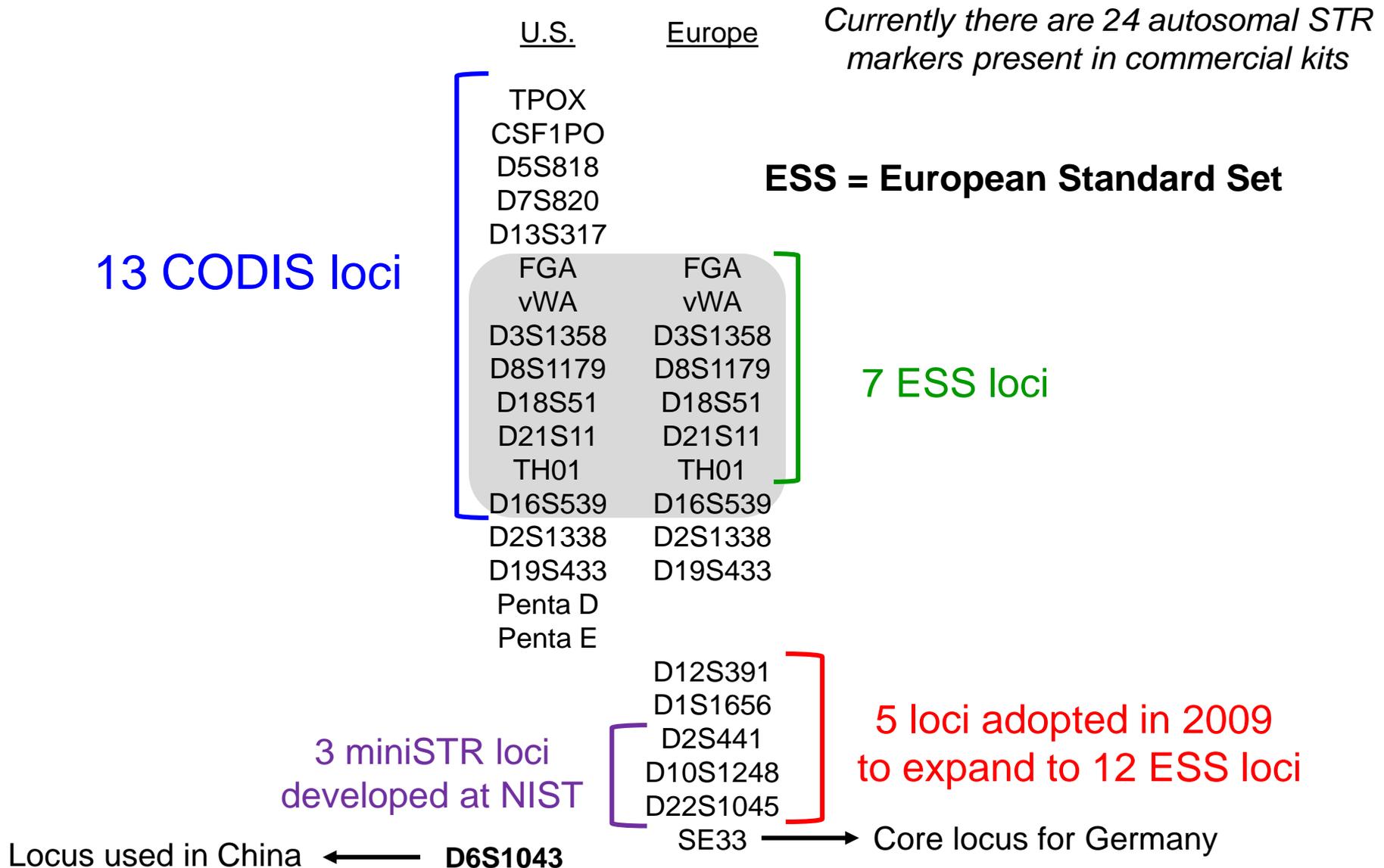
NIST



Standard Approaches Enable Reliable DNA Data Comparison

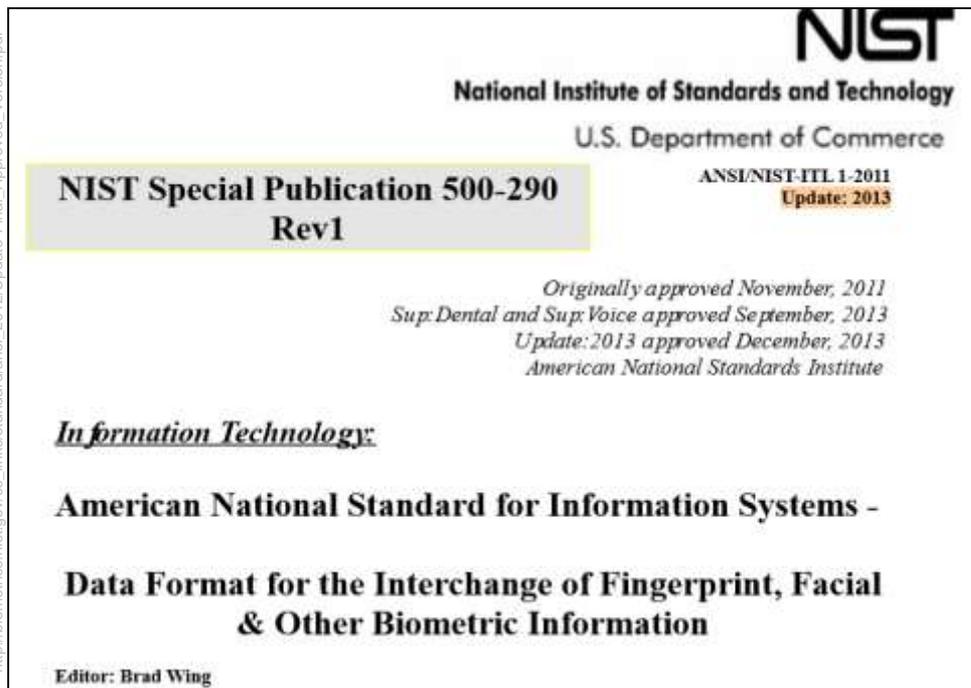
- **Core loci**
 - In 1997, U.S. selected 13 core STR markers
 - U.S. is moving to 20 core STRs in the near future
 - Europe moved from 7 to 12 core STR loci in 2011
- **Common data formats**
 - ISFG DNA Commission allele nomenclature designation recommendations
 - ANSI/NIST-ITL standard for data storage and transmission
- **Commercial STR kits**
 - Consistent allelic ladders
- **Certified reference materials**
 - NIST SRM 2391c (certified values for STR allele measurements)

International Comparability



ANSI/NIST-ITL Standard Data Format

http://www.nist.gov/itl/iad/ig/ansi_standard.cfm



623 page document

Latest update: December 2013

- **Data storage and transmission standard for software developers**
- Record types include biometric fingerprint, iris, dental, and voice information
- **DNA records (type 18) are covered in 24 pages**
- Provides list and codes for **88 DNA kits** from Life Technologies, Promega, and Qiagen
- Codes provided for **64 autosomal STR loci, 64 X-STRs, and 135 Y-STRs**

http://biometrics.nist.gov/cs_links/standard/ansi_2012/Type_18_DNA_Record_Kits_List_111913.pdf

http://biometrics.nist.gov/cs_links/standard/ansi_2012/Type_18_DNA_Record_Loci_list_111913.pdf

Commercially Available STR Kits

Applied Biosystems (17)

- ~~AmpFISTR Blue (1996)~~
- ~~AmpFISTR Green I (1997)~~
- Profiler (1997)
- Profiler Plus (1997)
- COfiler (1998)
- SGM Plus (1999)
- **Identifiler** (2001)
- Profiler Plus ID (2001)
- ~~SEfiler (2002)~~
- **Yfiler (2004)**
- MiniFiler (2007)
- SEfiler Plus (2007)
- Sinofiler (2008) – China only
- **Identifiler Direct** (2009)
- NGM (2009)
- **Identifiler Plus** (2010)
- NGM SElect (2010)
- GlobalFiler Express (2012)
- GlobalFiler Casework (2013)
- **Yfiler Plus (2014)**

Promega Corporation (18)

- PowerPlex 1.1 (1997)
- PowerPlex 1.2 (1998)
- PowerPlex 2.1 (1999)
- **PowerPlex 16** (2000)
- PowerPlex ES (2002)
- **PowerPlex Y (2003)**
- PowerPlex S5 (2007)
- **PowerPlex 16 HS** (2009)
- PowerPlex ESX 16 (2009)
- PowerPlex ESX 17 (2009)
- PowerPlex ESI 16 (2009)
- PowerPlex ESI 17 (2009)
- PowerPlex 18D (2011)
- PowerPlex 21 (2012)
- PowerPlex ESI 17 Pro (2012)
- PowerPlex Fusion (2012)
- **PowerPlex Y23 (2012)**
- PowerPlex Fusion 6C (2014)

Qiagen (2010)

Selling kits in Europe

*Due to patent restrictions
cannot sell in U.S.
until June 2015*

- ESSplex
- ESSplex SE
- Decaplex SE
- IDplex
- Nonaplex ESS
- Hexaplex ESS
- HD (Chimera)
- Argus X-12
- **Argus Y-12**
- **DIplex (30 InDels)**
- 24plex

Why do U.S. forensic DNA labs care about the use of reference materials?



9.5.5. The laboratory shall check its DNA procedures annually or whenever substantial changes are made to a procedure against an appropriate and available NIST standard reference material or standard traceable to a NIST standard.

Provides calibration and traceability

Reference Materials for Forensic DNA

Produced by the NIST Applied Genetics Group

NIST Standard Reference Material	Release date and renewals	Purpose/contents
SRM 2390	1992, 1999, 2001	DNA profiling standard (RFLP) <i>(discontinued in 2005)</i>
SRM 2391c	1995, 1999, 2002, 2008, 2011, 2014	PCR-based DNA profiling standard (autosomal STRs; now Y-STRs since 2011)
SRM 2392-I	2003, 2009, 2012	Mitochondrial DNA sequencing (human HL-60 DNA)
SRM 2395	2003, 2008	Human Y-chromosome DNA profiling standard <i>(discontinued in 2013)</i>
SRM 2372	2007, 2013	Human genomic DNA quantitation standard

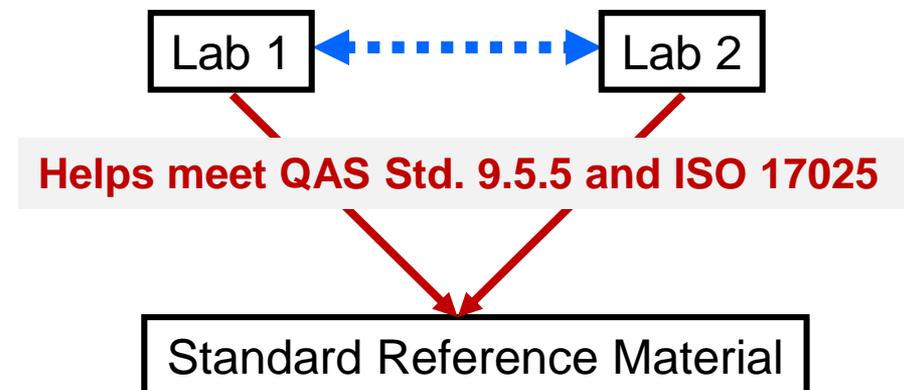
See <http://www.nist.gov/srm/>

NIST Standard Reference Material (SRM) 2391c: PCR-Based DNA Profiling Standard

- Components A through D are DNA extracts in liquid form
- Components E and F are DNA spotted on 903 paper or FTA paper
- **Certified values** are for STR alleles based on length polymorphisms observed using capillary electrophoresis



Genomic DNAs characterized for the expanded CODIS core loci and Y-STRs



Calibration with SRMs enables confidence in comparisons of results between laboratories

Standard Information Resources

NIST STRBase website: <http://www.cstl.nist.gov/strbase/>



Short Tandem Repeat DNA

Internet DataBase



NIST Standard Reference Database SRD 130

[Recent Updates]

Serving the forensic DNA and human identity testing communities for over 10 years... These data are intended to benefit research and application of short tandem repeat DNA markers to human identity testing. The authors are solely responsible for the information herein. **Please Rate Our Products and Services:** <http://tsapps.nist.gov/MSDSurvey/default.aspx?ID=5&DB=130>

This database has been accessed >500,000 times since 10/02/97.

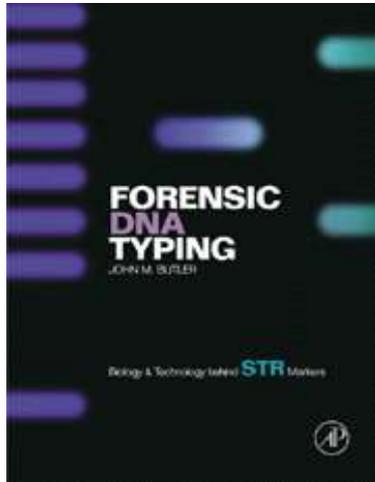
Created by John M. Butler

Forensic DNA Typing Textbooks Have Set the Standard for the Field

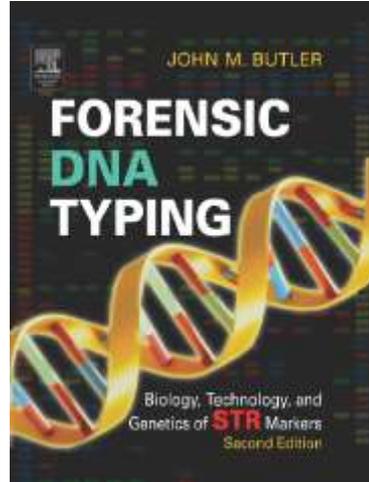
1st Edition

2nd Edition

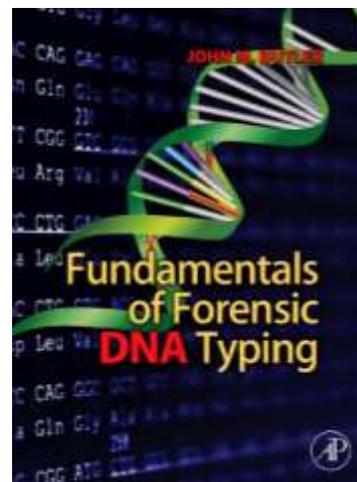
3rd Edition (3 volumes)



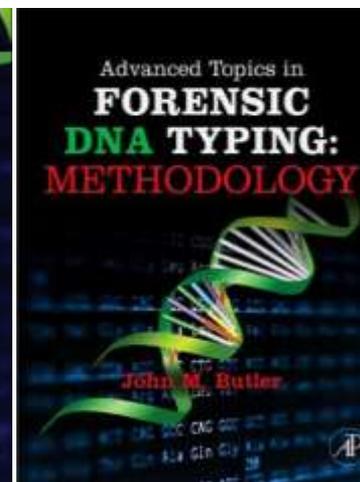
Jan 2001
335 pages



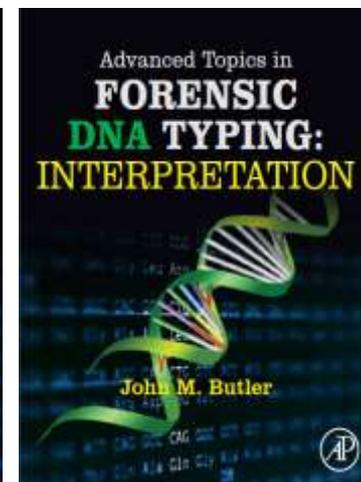
Feb 2005
688 pages



Sept 2009
520 pages



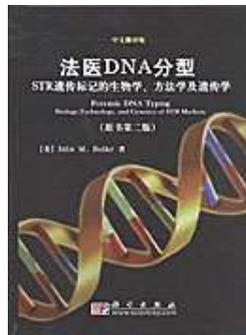
Aug 2011
704 pages



Sept 2014
604 pages

Language Editions

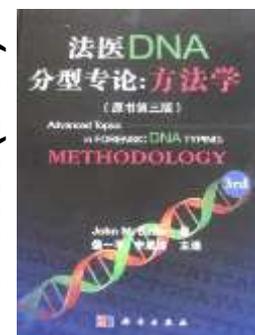
Chinese (2007)



Japanese (2009)

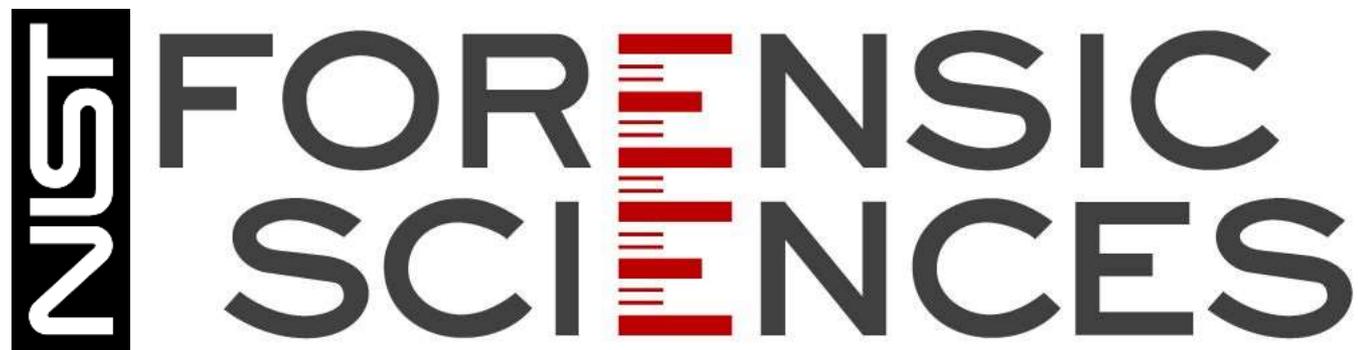


Chinese (2013)

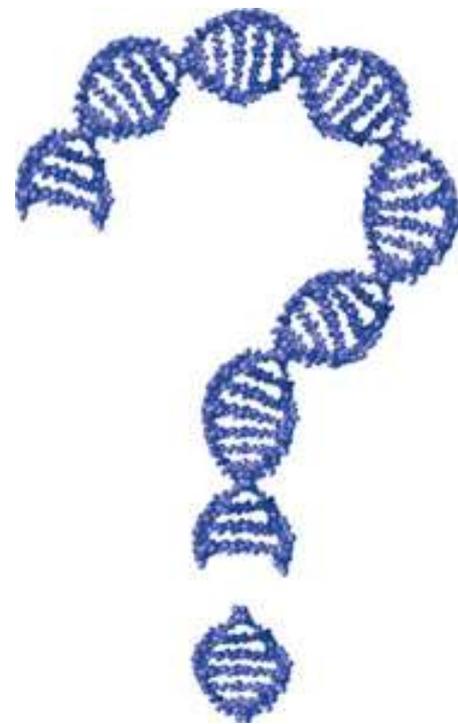


National Commission on Forensic Science (NCFS):
www.justice.gov/ncfs

Organization of Scientific Area Committees (OSAC):
www.nist.gov/forensics/osac/index.cfm



www.nist.gov/forensics



+1-301-975-4049

john.butler@nist.gov